A METHOD FOR DETECTING DISEASE-ASSOCIATED MUTATIONS

ABSTRACT

A method is described for diagnosing individuals as having hypertrophic cardiomyopathy, e.g. familial or sporadic hypertrophic cardiomyopathy. The method provides a useful diagnostic tool which becomes particularly important when testing asymptomatic individuals suspected of having the disease. Symptomatic individuals have a much better chance of being diagnosed properly by a physician. Asymptomatic individuals from families having a history of familial hypertrophic cardiomyopathy may be selectively screened using the method of this invention allowing for a diagnosis prior to the appearance of any symptoms. Individuals having the mutation responsible for the disease may be counseled to take steps which hopefully would prolong their life, i.e. avoid rigorous exercise. The methodology used in the above method also has broad applicability and may be used to detect other disease-associated mutations in DNA obtained from subjects being tested for other disease-associated mutations.